a guide for parents-to-be

nest

non-invasive prenatal screening tests

What is a nest prenatal screening test?

nest is a simple blood test that screens for chromosomal conditions that can affect your baby's health. A blood sample can be drawn at any of our specialised collection centres as early as the 10th week of pregnancy. You have two non-invasive prenatal screening options with nest; the standard **nest** test and the more comprehensive **nest+** test*. Test results are usually reported back to your doctor within 5 days from receipt in the lab.

Visit nestscreen.com.au to find your nearest collection centres.

Is nest right for me?

nest offers parents-to-be a reliable choice to obtain important information about the health of their developing baby, simply, accurately and in the first trimester (from 10 weeks), with no risk to their pregnancy.

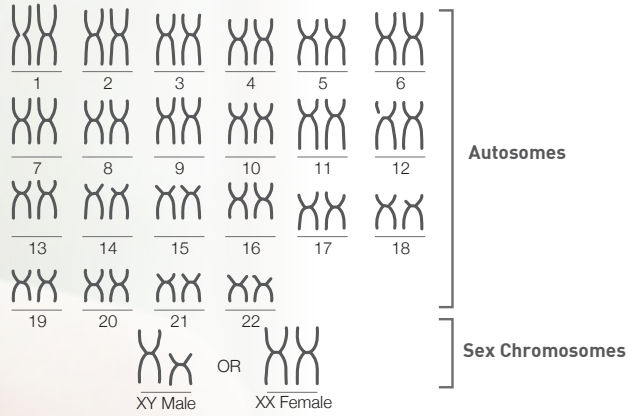
How does the test work?

A sample of your blood is drawn and the genetic material (DNA) from you and your baby is tested. The **nest** screening tests are precise methods that use an advanced technology called 'massively parallel sequencing' to analyse millions of DNA fragments per sample and accurately count the number of chromosomes present. We can then determine if your pregnancy has too many or too few copies of these chromosomes. Conditions with an extra chromosome are a trisomy, one less is a monosomy.

What specific conditions can nest screen for?

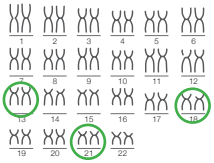
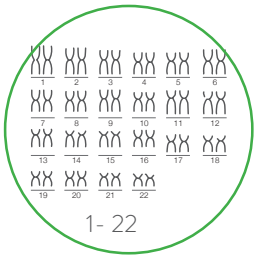
Chromosomes normally come in pairs. Most individuals have 23 pairs of chromosomes made up of 22 autosomes and a pair of sex chromosomes. Missing or extra copies of chromosomes can be associated with intellectual or physical disabilities, with differing levels of severity.

The chromosome make-up of an individual human cell



*nest+ is available for singleton pregnancies only.

Test comparison

	nest	nest+
Is the test suitable for a twin pregnancy?	Yes	No
What chromosomes are routinely screened? ◯ = Chromosomes tested	 13, 18, 21 Patau, Edwards and Down syndrome	 1- 22 Patau, Edwards and Down syndrome & Rare Autosomal Trisomies
Can Sex Chromosomes be screened?	Yes (singleton only)	Yes (singleton only)
Can I find out my baby's gender?	Yes	Yes
How early in my pregnancy can I do the test?	From 10 weeks gestation onwards	From 10 weeks gestation onwards
How much blood is collected?	A single 7 mL tube of blood	A single 7 mL tube of blood
How long does it take for my results to be reported?	Within 5 business days from receipt in lab	Within 5 business days from receipt in lab
Will my test fail? Will I get a result?	**nest** has an exceptionally low failure rate (0.1%)[1]. This means you are highly unlikely to have delayed results	**nest+** has an exceptionally low failure rate (0.1%)[1]. This means you are highly unlikely to have delayed results
Is a nest prenatal screening test cost covered by Medicare?	No, there is no Medicare rebate for anynon-invasive prenatal test in Australia.	No, there is no Medicare rebate for any non-invasive prenatal test in Australia.

The most commonly seen chromosomal conditions include;

- Trisomy 13 (Patau syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 21 (Down syndrome)

All of these conditions can be accurately detected with the nest prenatal screening tests.

If your doctor chooses to further personalise your screening, they may request nest to screen for the following sex chromosome conditions,

- Turner syndrome (only one X chromosome in a female)
- Klinefelter syndrome (an extra X chromosome in a male)
- Triple X syndrome (an extra X chromosome in a female)
- Jacobs syndrome (an extra Y chromosome in a male)

A Trisomy involving a chromosome other than 13, 18, 21, X or Y is referred to as a Rare Autosomal Trisomy (RAT).

Your doctor may choose to screen for these RATs and request the comprehensive nest+ prenatal screening test.

Although “rare” the prevalence of RATs detected by tests such as nest+ is significant, occurring in approximately 0.5 % of all pregnancies. The clinical outcome from a RAT is varied and chromosome dependant, outcomes may include pregnancy loss, fetal chromosomal abnormalities or confined placental mosaicsm (which could lead to uterine growth restrictions).

Your doctor may choose to request the comprehensive nest+ prenatal screening test. You can also elect to have gender reported to you.

What do my **nest** results mean?

Your results will tell your doctor whether or not Trisomies 13, 18, 21, Rare Autosomal Trisomies (nest+) or Sex chromosome conditions (if ordered) are likely to be present in your pregnancy. Your test report will include one of two possible results for the chromosomes screened,

- Low probability—means the expected number of chromosomes were found.
- High probability—means too many or too few copies of one of the chromosomes have been identified. This can indicate a chromosomal condition. Diagnostic testing is recommended for confirmation.

Each result will include an interpretation. If gender is requested this will be labelled male or female.

In the case of a high probability result, your doctor and genetic counsellor will discuss what the results mean to your pregnancy as well as further testing options to consider.

nest genetic counselling

Highly trained nest genetic counsellors are available at no cost if requested by your doctor. A genetic counsellor can explain this technology and how the results will be reported. They may also deliver your results if requested. Having a deep understanding of genetics and non- invasive prenatal screening tests gives them the skills to interpret these results and assist in explaining them. If your results do come back showing a high probability, the genetic counsellor can assist in explaining the next steps.

Do **nest** results mean that my baby will be perfectly healthy?

nest prenatal screening tests are highly accurate advanced screening tests that are non-invasive. No test, however, can guarantee a baby will not have any medical issues.

The standard **nest** test only screens autosomes 13, 18, 21, and sex chromosomes*, if ordered.

The comprehensive **nest+**** test screens all 22 autosomes (including 13,18 and 21) and sex chromosomes*, if ordered.

The nest prenatal screening tests do not test for or report on all genetic and non-genetic problems that may be present in a baby.

Genetic counselling is available to all patients on request to assist in understanding what to expect from their results.

Women who receive high probability results should undertake invasive prenatal procedures for confirmation.

If the test result is low probability, it does not completely rule out all potential problems associated with chromosomes, or all sex chromosome conditions in your baby.

A low probability test result does not ensure an unaffected pregnancy. CVS and amniocentesis provide definitive diagnostic information, but the invasive nature of these procedures means that they are generally only offered to those who have been identified as a high risk pregnancy.

Early non-invasive prenatal screening tests such as nest are an important part of modern obstetric care alongside high quality ultrasound.

When considering the suitability of this test for your pregnancy please consult your Doctor.

*sex chromosome aneuploidy testing is available for singleton pregnancies only

****nest+*** non invasive prenatal test is available for singleton pregnancies only.

nest notes

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To learn more abut **nest** please visit **nestscreen.com.au**
or phone **1800 874 971**

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References
1. Bhatt S, Parsa S, Snyder H, Taneja P, Halks-Miller M, Seltzer W, DeFeo E. Clinical Laboratory Experience with Noninvasive Prenatal Testing: Update on Clinically Relevant Metrics. ISPD 2014 poster.
2. Bianchi DW, Platt LD, Goldberg JD, et al. Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. Obstet Gynecol. 2012;119:890–901.
3. Futch T, Spinosa J, Bhatt S, de Feo E, Rava RP, Sehnert AJ. Initial clinical laboratory experience in noninvasive prenatal testing for fetal aneuploidy from maternal plasma DNA samples. Prenat Diagn. 2013;33:569-574.